Appendix 1

Glossary of Selected Genetic Terms

Association: two or more malformations, which have been found (on a population basis) to occur together more often than would be predicted by chance, but not necessarily due to a specific causal entity; examples: VATER association, consisting of **V**ertebral, **A**nal, **T**racheal, **E**sophageal and **R**adial or **R**enal defects; CHARGE association, consisting of **C**oloboma, **H**eart, **A**tresia choanae, **R**etardation of growth and development, **G**enital and **E**ar defects

Biochemical analysis: qualitative or quantitative analysis of amino acids, organic acids, proteins, enzymes or other components, usually performed in order to evaluate the possibility of an inborn error of metabolism

Chromosomal analysis (karyotype), routine: cytogenetic analysis to determine the number and structure of chromosomes as seen through microscopic views of prepared cells from an individual's tissue; the typical process involves *in vitro* culture, fixation and staining; counting and analysis of the chromosomal number and structure of representative cells; and preparation of photographs of representative cells with a written interpretation of the findings (see Giemsa banding)

Chromosomal analysis, high resolution: cytogenetic laboratory method in which the chromosomes are induced to be more elongated, allowing much finer detection of smaller changes in the pattern of banding than usual; also called high resolution banding or prometaphase banding

Clinical geneticist: a physician with special training in the clinical evaluation of patients suspected to have genetic conditions and birth defects; clinical geneticists historically came from a variety of backgrounds, such as pediatrics, obstetrics/gynecology, internal medicine, neurology, etc. but newly trained individuals may specialize in genetics alone

Congenital anomaly: a structural abnormality present at birth; also termed a malformation; examples: cardiac ventricular septal defect or microphthalmia

Congenital: present at birth, although not necessarily detected at that time; for example, a Meckel's diverticulum may not be detected until adulthood

Consanguinity: sharing of a common blood relative by two individuals contemplating parenthood together or already parents; individuals who are first cousins share 1/8 of their genes in common, which increases the likelihood that a recessive genetic change would be shared by both

Cytogenetic: referring to the analysis of chromosomes within cells, by culturing living cells or preparing actively dividing cells from body tissues and use of various identification methods to detect individual chromosome structure

Deformation: a congenital structural variation or abnormality which is the result of external physical forces, such as constraint within the uterine cavity; some self correct with time; others may respond to physical stretching treatments; example: club foot which responds to casting

Developmental variant or variation: a cosmetically and functionally insignificant structural deviation, often familial and of prenatal origin

Disruption: a congenital abnormality which is the result of a destructive event or process, leading to incomplete or abnormal formation; example: failure of vascular flow to portions of the cerebrum, resulting in porencephaly (a vascular disruption) or absence of toes due to amniotic bands

Dysmorphic: literally, abnormally shaped; refers to a physical feature which is sufficiently different from average as to cause concern

Dysmorphologist: a specialist trained in the recognition of physical variations and malformations and their diagnostic and clinical significance

Fluorescence in situ hybridization (FISH): a molecular cytogenetic method in which a molecular probe is linked to a fluorescent dye; after preparation of a cytogenetic or cellular specimen the probe is placed on the slide; views under a fluorescent microscope reveal presence or absence of the desired region of the chromosome through adhesion of the probe or lack thereof

Fragile X analysis: methods for detecting the presence of the fragile site at Xq27.3; historically, this was performed by cytogenetic analysis of cells grown in a special growth medium but this testing has been replaced by molecular methods to detect the number of CGG repeats in the chromosome region, with defined ranges for normal and abnormal results

Genetic counseling: a process by which individuals or families receive information about genetic disorders or malformations and risks for these conditions after review of family and medical history; includes analysis of diagnostic information, review of natural history of the relevant conditions, options for dealing with risks and enhancement of family decision-making through non-judgmental support (see Counseling the Family)

Genetic counselor: a professional trained in counseling families about genetic conditions, genetic risks and decision-making and adapting to a genetic condition

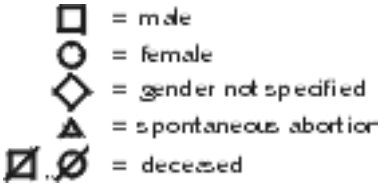
Genetic support group: a group or organization developed around a specific condition or group of conditions related to genetics or malformations, in order to provide information and support to families

Genetic discrimination: use of information about genetic traits in order to identify at-risk individuals for a different selection process in employment, housing, insurance or other usually non-medical settings; typically used to refer to an *unfair* selection process by which individuals are denied or presented with barriers to their application, such as much higher premiums for insurance

Giemsa banding: a laboratory method for precisely identifying chromosomes, by means of a pattern of microscopic dark and light bands; both large and moderate gains or losses of chromosomal material can be detected

Hereditary: inherited or inheritable through the gametes of biological parents; commonly refers to traits related to a single gene or specific chromosome alteration

Key to Symbols for Pedigree Diagrams:



Major malformation or anomaly: a congenital structural abnormality which has significant effect on function or social acceptability; example: cleft lip; in its strictest definition, *malformation* defines a structural abnormality resulting from an abnormal developmental process, but common usage includes all structural abnormalities as malformations, regardless of cause (see *Disruption* and *Deformation*)

Medical geneticist: a professional with a doctoral degree and special training in the relationships between genes and disease (see *Clinical geneticist*)

Mendelian inheritance: pattern of inheritance of a genetic trait due to a single gene variation which follows the classical patterns of autosomal recessive, autosomal dominant or X-linked (dominant or recessive) inheritance

Metabolic analysis: same as biochemical analysis but also includes many commonly performed blood chemistries used to detect an inborn error of metabolism, such as ceruloplasmin or cholesterol

Microdeletion syndrome: pattern of malformations due to loss of a chromosomal segment containing several consecutive (contiguous) genes; they may be difficult or impossible to detect by routine cytogenetic analysis and usually require high resolution (> 550 band) analysis, a molecular test or a molecular cytogenetic test (FISH) for confirmation; example: Prader-Willi syndrome

Minor malformation: congenital structural abnormality which has little functional or societal significance on its own; example: preauricular pit, syndactyly of the second and third toes

Molecular (DNA) test: laboratory analysis of an individual's genetic material using a designated piece of DNA coding for a region or gene of interest (a probe), in which variations within the region or gene are detected by adherence or lack of adherence of the probe to the corresponding piece of DNA from the individual; the probe is labeled by fluorescent dyes, radioactive labeling or other methods; see *Table 6* for a list of some available molecular (DNA) tests

Molecular cytogenetic test: laboratory analysis of an individual's body cells using a combination of cytogenetic analysis to view chromosomes and a molecular test with detectable (usually fluorescent) probes which allows the presence or absence of the region(s) of interest to be recognized

Monogenic: due to a single gene

Natural history: typical course of a medical condition in the absence of specific intervention

Pedigree: a formal diagram of the blood relationships in a family using standardized symbols and nomenclature, indicating specific medical conditions, the status of each family member in relation to pertinent genetic traits (affected, unaffected, carrier, etc.) and additional relevant information necessary to discern possible patterns of inheritance and genetic risk; typically three or more generations are recorded

Phenotype: the clinical or behavioral presentation of a genetic variation in an individual

Phenotype, abnormal: the clinical presentation of a genetic or malformation disorder in an individual, including associated complications and their severity

Phenotype, behavioral: the pattern of behavioral abnormalities and traits associated with a specific genetic or malformation disorder

Pierre Robin sequence: a triad of congenital micrognathia (small mandible), retropositioning of the tongue (glossoptosis) and upper airway obstruction, usually with a U-shaped palatal cleft; a component malformation of a number of distinct syndromes

Polydactyly: extra fingers or toes; may be isolated or may be a component of malformation syndromes

Recurrence risk: the likelihood that a clinical disorder will occur in additional siblings of the same biological parents

Sequence: a pattern of related malformations and/or deformations and/or disruptions such that one initial physical abnormality leads to the occurrence of additional abnormalities in a sequential manner; example: meningomyelocele leading to paralysis of lower limbs leading to club feet

Syndrome: a recognizable recurrent pattern of malformations and/or deformations and/or disruptions with one or more specific, defined causes; examples: Prader-Willi syndrome due to 15q deletion, Fetal alcohol syndrome due to prenatal ethanol exposure, Jeune syndrome due to an as-yet unidentified recessive gene

Teratogen: a biochemical or other agent capable of inducing one or more malformations in a developing embryo

Appendix 2

Selected Reference Charts for Newborns

2A. Gestational Age Assessment (Ross Laboratories, with permission)

GESTATIONAL AGE ASSESSMENT (Dubowitz)

NAME _____ **DATE/TIME OF BIRTH** _____ **BIRTH WEIGHT** _____

HOSPITAL NO. _____ **DATE/TIME OF EXAM** _____ **LENGTH** _____

AGE WHEN EXAMINED _____ **HEAD CIRCUMFERENCE** _____

RACE _____ **SEX** _____ **EXAMINER** _____

APGAR SCORE: 1 MINUTE _____ **5 MINUTES** _____

NEUROLOGICAL CRITERIA

NEUROLOGICAL SIGN	SCORE 0	1	2	3	4	5	RECORD SCORE HERE
POSTURE							
SQUARE WINDOW	 90°	 60°	 45°	 30°	 0°		
ANKLE DORSI-FLEXION	 90°	 75°	 45°	 20°	 0°		
ARM RECOIL	 180°	 90-180°	 <90°				
LEG RECOIL	 180°	 90-180°	 <90°				
POPLITEAL ANGLE	 180°	 160°	 130°	 110°	 90°	 <90°	
HEEL TO EAR							
SCARF SIGN							
HEAD LAG							
VENTRAL SUSPENSION							
						TOTAL NEUROLOGICAL SCORE	

EXTERNAL (SUPERFICIAL) CRITERIA

EXTERNAL SIGN	SCORE 0	1	2	3	4	RECORD SCORE HERE
EDEMA	Obvious edema of hands and feet; pitting over tibia	No obvious edema of hands and feet; pitting over tibia	No edema			
SKIN TEXTURE	Very thin, gelatinous	Thin and smooth	Smooth; medium thickness. Rash or superficial peeling	Slight thickening. Superficial cracking and peeling, esp. hands, feet	Thick and parchment like; superficial or deep cracking	
SKIN COLOR (Infant not crying)	Dark red	Uniformly pink	Pale pink; variable over body	Pale. Only pink over ears, lips, palms, or soles		
SKIN OPACITY (Trunk)	Numerous veins and venules clearly seen, esp. over abdomen	Veins and tributaries seen	A few large vessels clearly seen over abdomen	A few large vessels seen indistinctly over abdomen	No blood vessels seen	
LANUGO (Over back)	No lanugo	Abundant; long and thick over whole back	Hair thinning, esp. over lower back	Small amount of lanugo and bald areas	At least half of back devoid of lanugo	
PLANTAR CREASES	No skin creases	Faint red marks over anterior half of sole	Definite red marks over more than anterior half; indentations over less than anterior third	Indentations over more than anterior third	Definite deep indentations over more than anterior third	
NIPPLE FORMATION	Nipple barely visible; no areola	Nipple well-defined; areola smooth and flat; diameter <0.75 cm	Areola stippled, edge not raised; diameter <0.75 cm	Areola stippled, edge raised; diameter >0.75 cm		
BREAST SIZE	No breast tissue palpable	Breast tissue on one or both sides <0.5 cm diameter	Breast tissue both sides; one or both 0.5-1.0 cm	Breast tissue both sides; one or both >1 cm		
EAR FORM	Pinna flat and shapeless, or no incurving of edge	Incurving of part of edge of pinna	Partial incurving of whole of upper pinna	Well-defined incurving of whole of upper pinna		
EAR FIRMNESS	Pinna soft, easily folded, no recoil	Pinna soft, easily folded, slow recoil	Cartilage to edge of pinna, but soft in places, ready recoil	Pinna firm, cartilage to edge, instant recoil		
GENITALIA MALE: FEMALE (With hips half-abducted)	Neither testis in scrotum Labia majora widely separated, labia minora protruding	At least one testis high in scrotum Labia majora almost cover labia minora	At least one testis fully descended. Labia majora completely cover labia minora			
					TOTAL SUPERFICIAL SCORE	

References Dubowitz LMS, Dubowitz V, Goldberg C. Clinical assessment of gestational age in the newborn infant *J Pediatr* 77:1-10, 1970.
Dubowitz LMS, Dubowitz V: *Gestational Age of the Newborn*. Reading, Mass: Addison-Wesley Publishing Co., 1977.

SCORE

Neurological _____

Superficial _____

Total _____

Total Score	Gestational Age (weeks)
10	27.2
12	27.8
14	28.3
16	28.8
18	29.4
20	29.9
22	30.4
24	30.9
26	31.5
28	32.0
30	32.5
32	33.0
34	33.6
36	34.1
38	34.6
40	35.2
42	35.7
44	36.2
46	36.7
48	37.3
50	37.8
52	38.3
54	38.9
56	39.4
58	39.9
60	40.4
62	41.0
64	41.5
66	42.0
68	42.6

GESTATIONAL AGE (weeks)

By dates _____

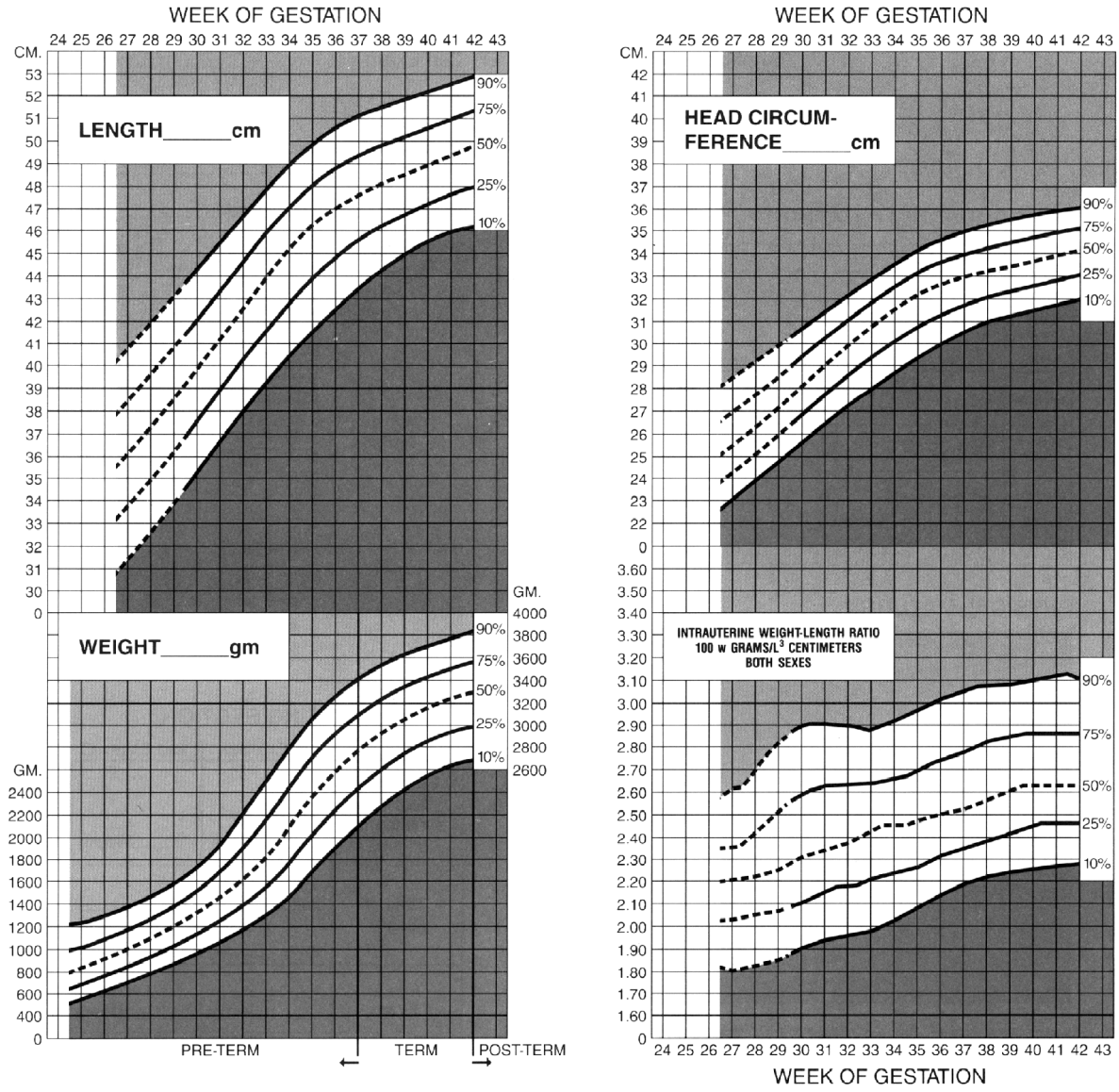
By ultrasound _____

By score _____

2B. Classification of Newborns: Intrauterine Growth (Mead Johnson Nutritionals, with permission)

CLASSIFICATION OF NEWBORNS –
BASED ON MATURITY AND INTRAUTERINE GROWTH
Symbols: X-1st Exam O-2nd Exam

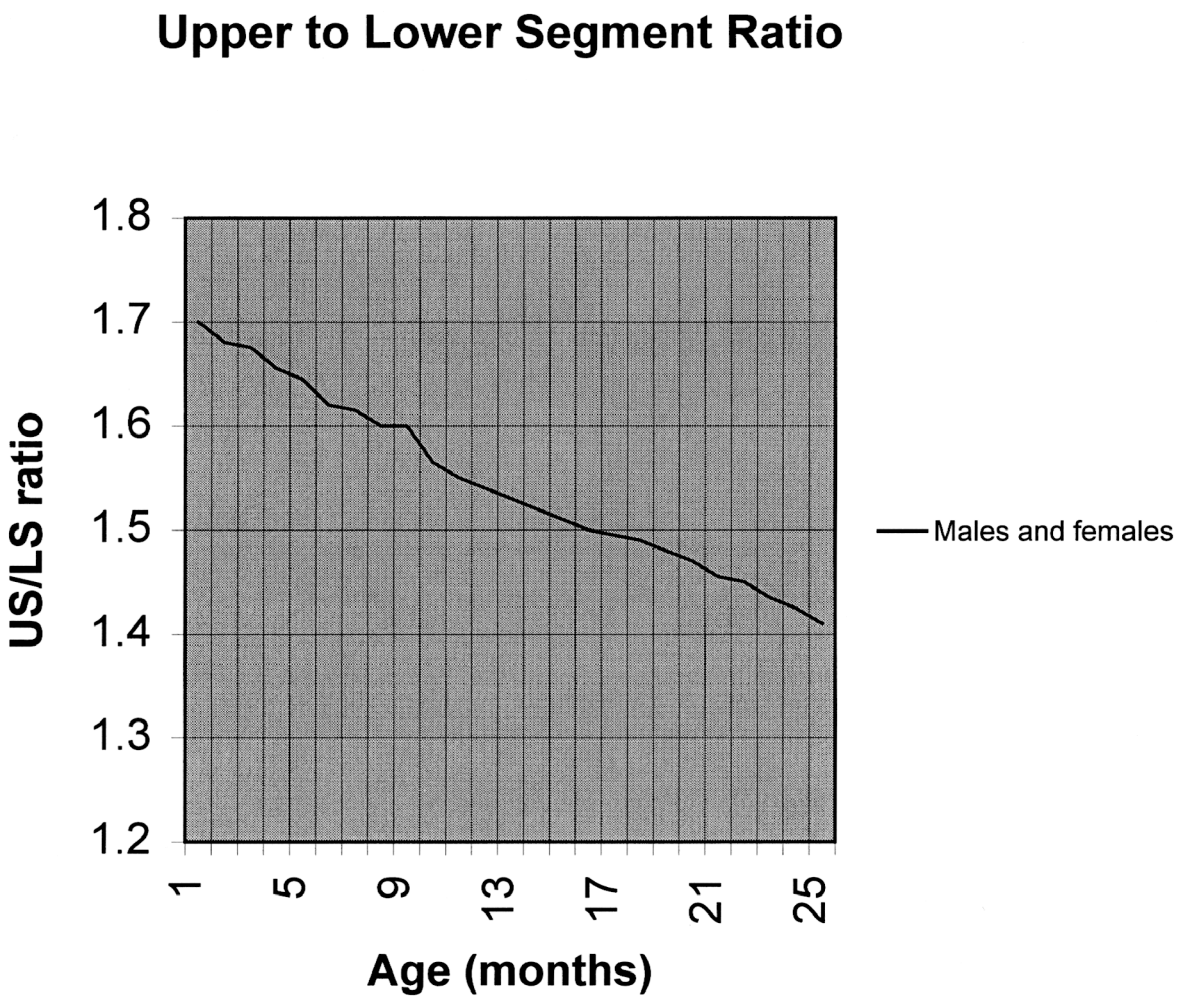
Side 2



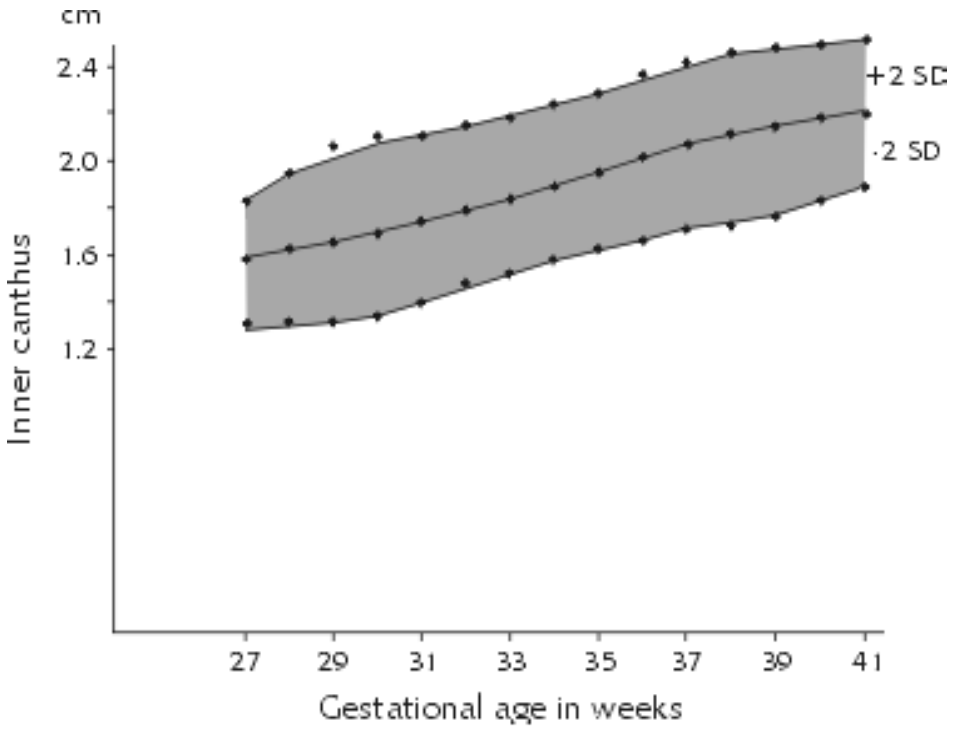
	1st Exam (X)	2nd Exam (O)
LARGE FOR GESTATIONAL AGE **(LGA)**		
APPROPRIATE FOR GESTATIONAL AGE **(AGA)**		
SMALL FOR GESTATIONAL AGE **(SGA)**		
Age at Exam	hrs	hrs
Signature of Examiner	M.D./R.N.	M.D./R.N.

Adapted from Lubchenco LO, Hansman C, and Boyd E: Pediatr. 1966; 37:403; Battaglia FC, and Lubchenco LO: J Pediatr. 1967; 71:159.

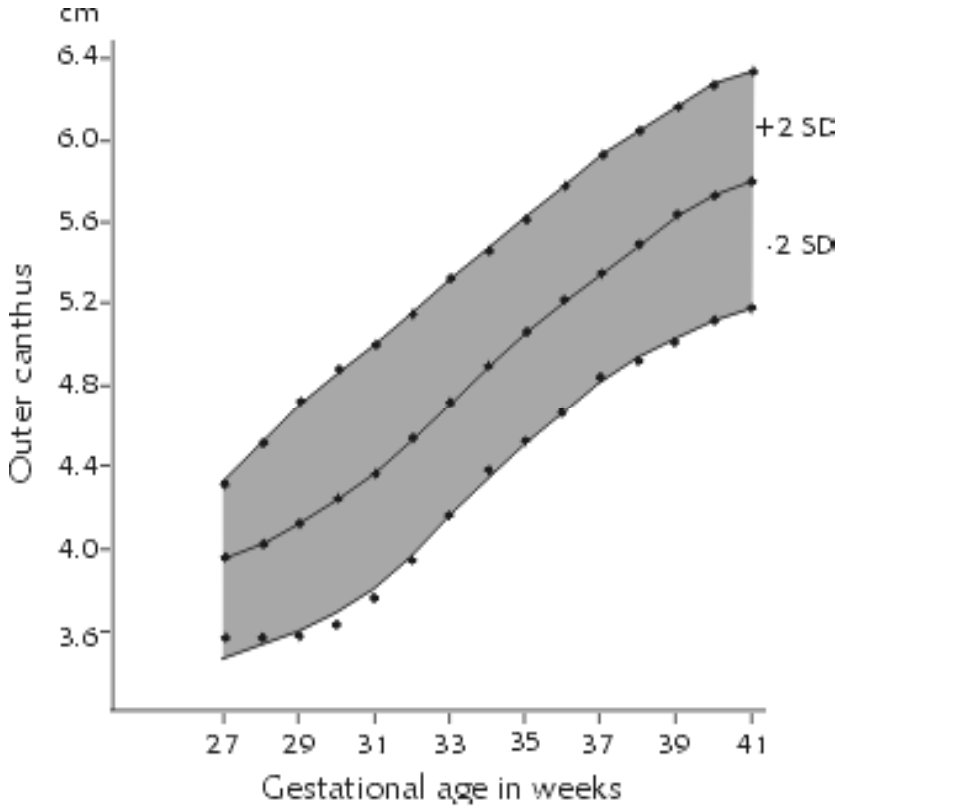
2C. Upper to Lower Segment Ratio (adapted from Harriet Lane Handbook, Mosby, Inc., with permission)



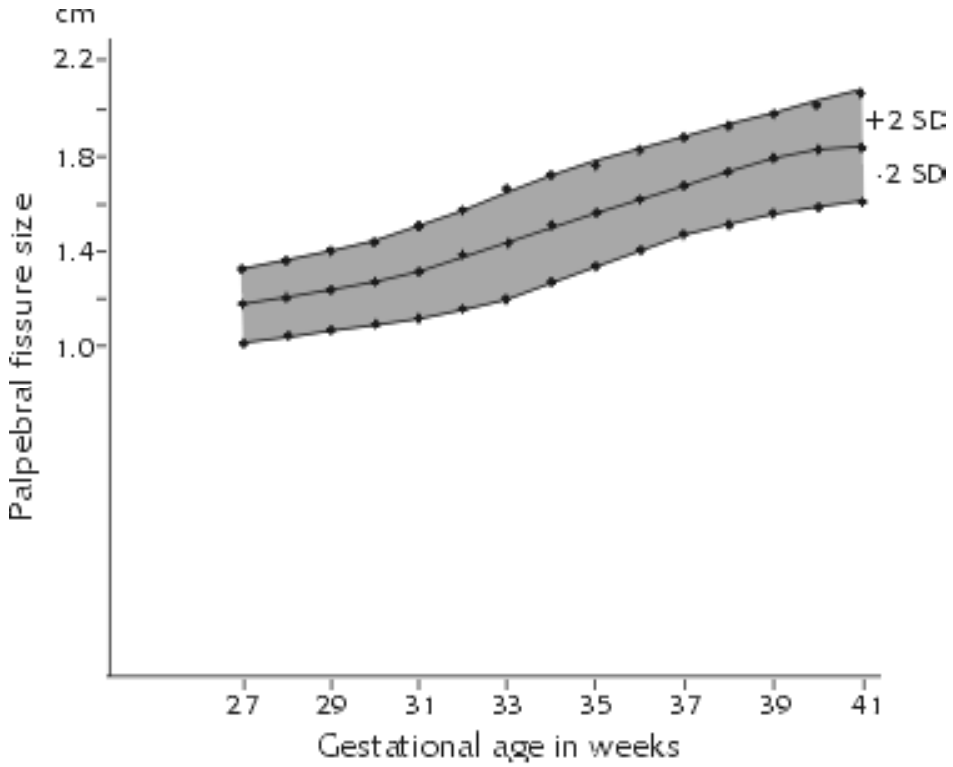
2D. Inner Canthal Measurement (Sivan et al, J Craniof Genet Devel Biol 2:239-242, 1982, with permission)



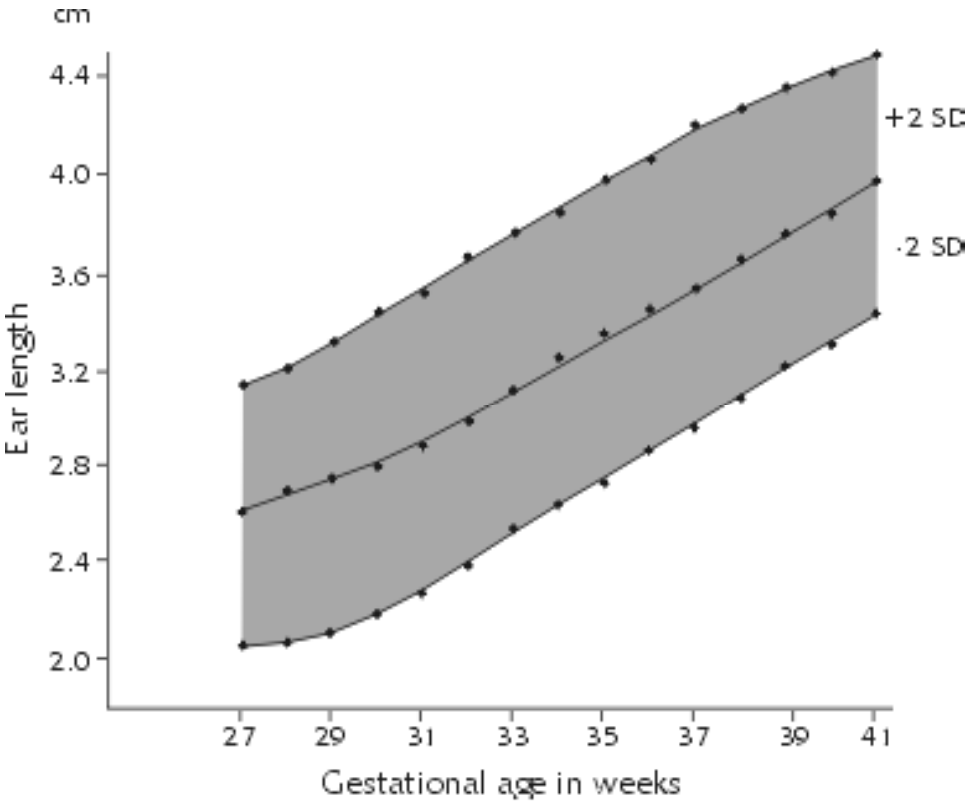
2E. Outer Canthal Measurement (Sivan et al, J Craniof Genet Devel Biol 2:239-242, 1982, with permission)



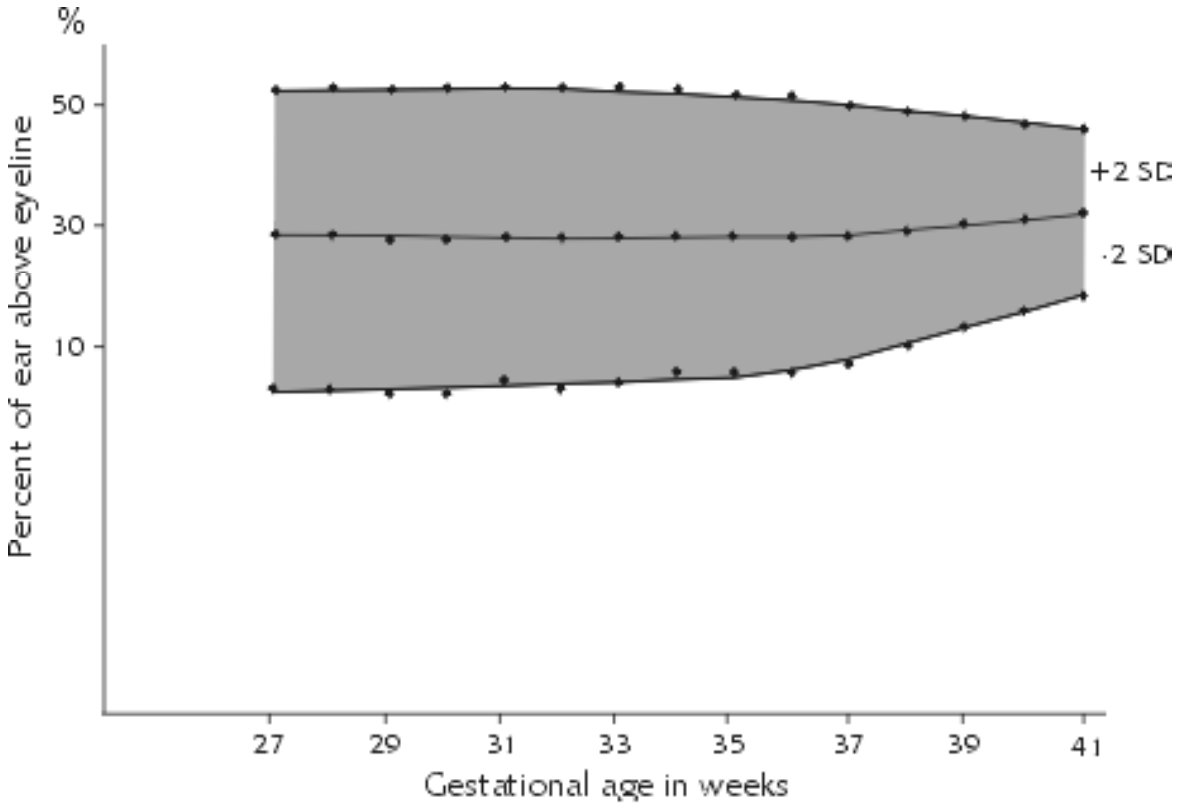
2F. Palpebral Fissure Length (Sivan et al, J Craniof Genet Devel Biol 2:239-242, 1982, with permission)



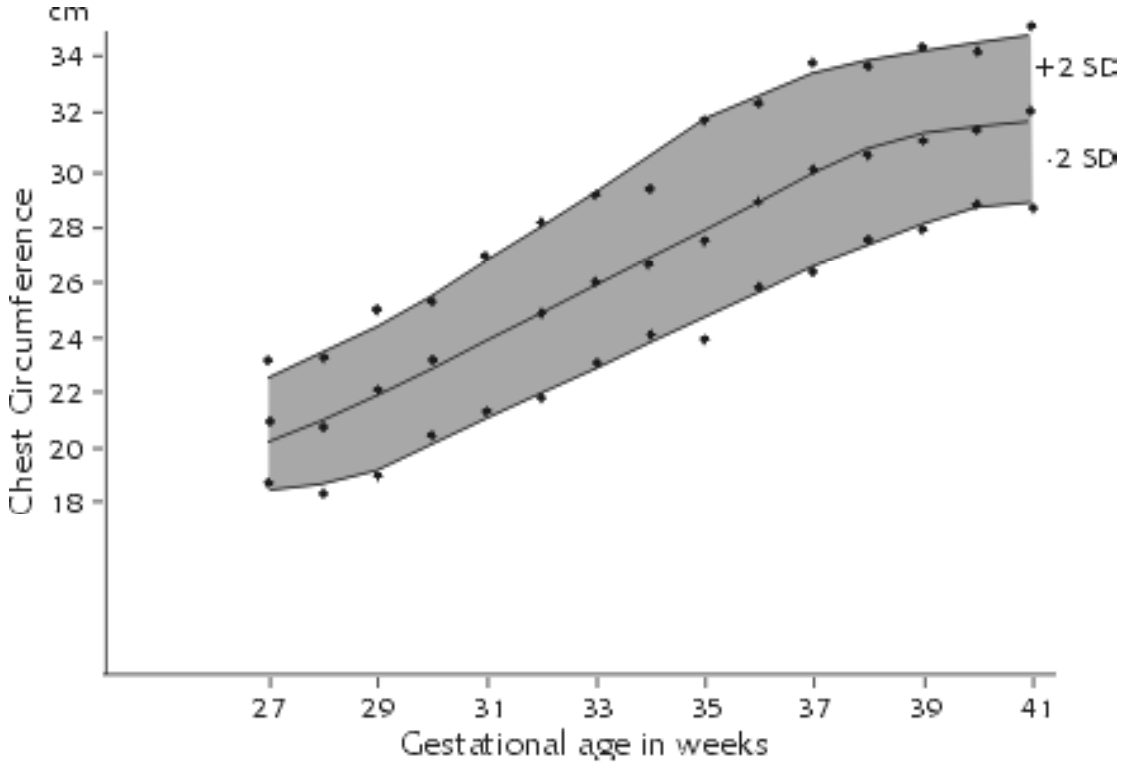
2G. Ear Length (J Med Genet 20(3): 213-215, 1983, with permission)



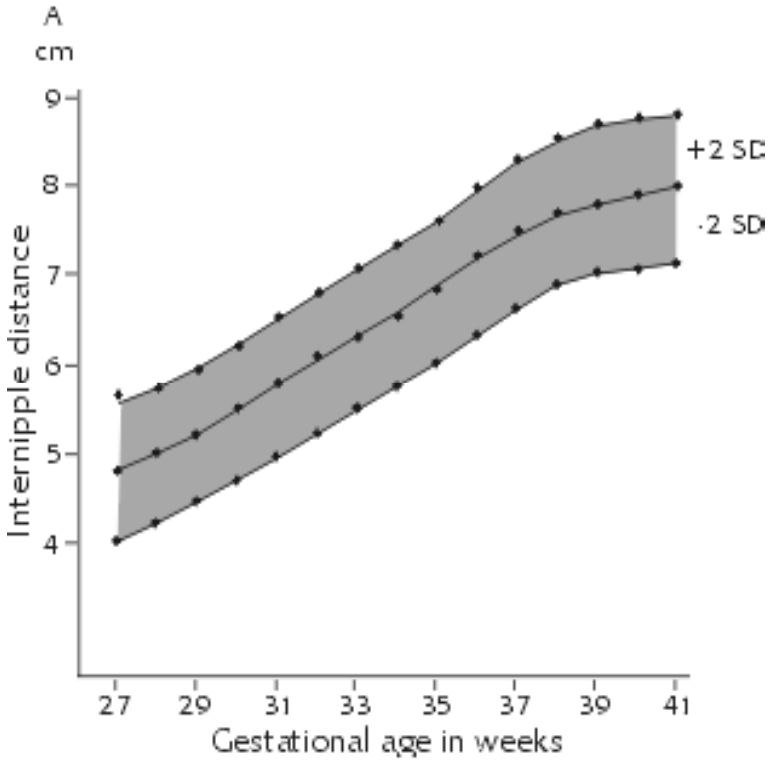
2H. Percent of Ear Above Eyeline (J Med Genet 20(3): 213-215, 1983, with permission)



2I. Chest Circumference (Merlob, March of Dimes Birth Defects Foundation, with permission)



2J. Internipple Distance (Sivan et al, Pediatr 72(4): 523-525, 1983, with permission)



Appendix 3

General Resources

Books

American Academy of Pediatrics. Genetic Disorders and Birth Defects: A Compendium of Guidelines and Resources for the Primary Care Practitioner. American Academy of Pediatrics, Elk Grove Village, 1997.

Buyse, M. Birth Defects Encyclopedia, 2 vols., Blackwell Scientific Publications, 1992.

Cohen MM Jr. The Child with Multiple Birth Defects, 2nd Edition. Oxford University Press, New York, 1997.

Gorlin RJ, Cohen MM, Levin LS. Syndromes of the Head and Neck, Third Edition. Oxford University Press, New York, 1990.

Gardner RJM, Sutherland GR. Chromosomal Abnormalities and Genetic Counseling. Oxford University Press, New York, 1989.

Graham, JM. Smith's Recognizable Patterns of Human Deformation. W. B. Saunders, Philadelphia, 1988.

Hall JG, Froster-Iskenius, Allanson JE. Handbook of Normal Physical Measurements. Oxford University Press, New York, 1989.

Seashore MR, Wappner RS. Genetics in Primary Care & Clinical Medicine. Appleton & Lange, Stamford, 1996.

Jones KL. Smith's Recognizable Patterns of Human Malformation, Fifth Edition. W.B. Saunders, Philadelphia, 1997.

McKusick VA. Mendelian Inheritance in Man, 11th Edition, Johns Hopkins University Press, Baltimore, 1994.

Rimoin D, Connor JM, Pyeritz RE. Emery and Rimoin's Principles and Practice of Medical Genetics, Third Edition. Churchill Livingstone, New York, 1996.

Robinson A, Linden MG. Clinical Genetics Handbook, Second Edition. Blackwell Scientific Publications, Boston, 1993.

Stevenson RE, Hall JG, Goodman RM. Human Malformations and Related Anomalies. Oxford University Press, New York, 1993.

Taybi H, Lachman R. Radiology of Syndromes, Metabolic Disorders and Skeletal Dysplasias, Fourth Edition. Mosby, St. Louis, 1996.

Thoene JG. Physician's Guide to Rare Diseases. Dowden Publishing Company, Montvale, NJ, 1992.

Wynne-Davies R, Hall CM, Apley AG. Atlas of Skeletal Dysplasias. Churchill Livingstone, London, 1985.

Zitelli BJ, Davis HW. Atlas of Pediatric Physical Diagnosis, Third Edition. CV Mosby, St. Louis, 1997.

On-Line Resources for Health Care Providers

On-line Mendelian Inheritance in Man (OMIM) (on-Line version of McKusick's book)
< *http://www3.ncbi.nlm.nih.gov/Omim/*>

Medical Search engines
Medical Matrix <*http://www.slackinc.com/matrix/*>
Cliniweb <*http://www.ohsu.edu/cliniweb/*>
PEDINFO <*http://www.uab.edu.pedinfo/*>

University of Kansas Genetics Professional Home Page (many links to other sources on genetics and congenital malformations)
<*http://www.kumc.edu/gec/geninfo.html*>

University of Kansas Support Groups Information and links
<*http://www.kumc.edu/gec/support/supgroup.html*>

Blazing a Genetic Trail (tutorial on basic genetics)
<*http://www.hhmi.org/GeneticTrail/*>

Description of Approach to the Dysmorphic Child
<*http://www.kumc.edu/instruction/medicine/pedcard/genetics/genetics.html*>

Pedinfo links for Congenital Diseases
<*http://www/UAB.EDU/pedinfo/DiseasesCongenital.html*>

List of Biochemical Genetic Tests by Disease (details of biochemical genetic testing)
<*http://biochemgen.ucsd.edu/wbgtests/dz-tst.htm*>

Helix Directory of Medical Genetics Laboratories (password needed)
<*http://healthlinks.washington.edu/helix*>

Alliance of Genetic Support Groups
<*http://medhlp.netusa.net/www/agsg.htm*>

March of Dimes Birth Defects Foundation
<*http://www.modimes.org*>

National Organization for Rare Disorders
<*http://www.pcnet.com/~orphan*>

March of Dimes Resource page
<*http://www.modimes.org/rc/help.htm*>

March of Dimes Fact Sheets On-Line/NOAH
<*http://www.noah.cuny.edu/pregnancy/pregnancy.html#BIRTHDEFECTS and gene*>

Genetic support group links
<*http://members.aol.com/dnacutter/sgroup.htm*>

Guide to genetic resources on the Web
<*http://www.dml.georgetown.edu/%7Edavidsol/len.html*>

Appendix 4

New York State Genetic Services Directory - Listing of Program Directors

BUFFALO REGION

Richard W. Erbe, M.D.
Chief, Division of Genetics
Children's Hospital of Buffalo
Division of Genetics
219 Bryant Street
Buffalo, NY 14222
Telephone: (716) 878-7530
FAX: (716) 878-7980

ROCHESTER REGION

George B. Segel, M.D.
Director, Division of Pediatric Genetics
University of Rochester School of Medicine
Department of Pediatrics, Box 777
601 Elmwood Avenue
Rochester, NY 14642
Telephone: (716) 275-3304
FAX: (716) 273-1018

SYRACUSE REGION

Robert Silverman, M.D.
Director of Perinatal Center
SUNY Health Science Center at Syracuse
90 Presidential Plaza
UHCC Building
Syracuse, NY 13202
Telephone: (315) 464-4458
FAX: (315) 464-8366

Joe Hoo, M.D.
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SUNY Health Science Center at Syracuse
Regional Genetics Program
Room 1214 - WSK
750 East Adams Street
Syracuse, NY 13210
Telephone: (315) 464-5884
FAX: (315) 464-6557

BINGHAMTON REGION

Luba Djurdjinovic, M.S.
Project Director/Genetic Counselor
Binghamton Genetic Counseling Program
124 Front Street
Binghamton, NY 13905
Telephone: (607) 724-4308
FAX: (607) 724-8290

ALBANY REGION

Ian H. Porter, M.D.
Director, Clinical Genetics
Albany Medical Center
Department of Pediatrics - A88
47 New Scotland Avenue
Albany, NY 12208
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FAX: (518) 262-5924

Alan E. Guttmacher, M.D.
Director
Vermont Regional Genetics Center
1 Mill Street, Box B-10
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HUDSON VALLEY REGION

Anna Bogdanow, M.D.
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Blythedale Children's Hospital
Valhalla, NY 10595-1597
Telephone: (914) 592-7555
FAX: (914) 592-5844

Stirling Puck, M.D.
Martha Weinstein, M.D.
Constance Sandlin, M.D.
Medical Directors
Genzyme Genetics
100 Corporate Drive
Yonkers, NY 10701-6807
Telephone:
Laboratory (914) 969-3399
Services (914) 345-3255
FAX: (914) 345-3732

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(914) 997-6535 (office)
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FAX: (914) 347-4218

Peggy Blattner, M.S.
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Scarsdale, NY 10583
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NEW YORK CITY - THE BRONX

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Lincoln Medical and Mental Health Center
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FAX: (718) 579-4640

Susan J. Gross, M.D.
Robert Marion, M.D.
Harold M. Nitowsky, M.D.
Co-Directors
Montefiore Medical Center
Division of Reproductive Genetics
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Bronx, NY 10461
Telephone: (718) 405-8150
FAX: (718) 405-8154

NEW YORK CITY - BROOKLYN

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Jessica G. Davis, M.D.
Nelly Jouayed, M.D.
Clinical Geneticists
The Brooklyn Hospital Center
Reproductive Genetics/Pediatric Genetics
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Brooklyn, NY 11201
Telephone: (718) 250-8032
FAX: (718) 250-8660

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FAX: (718) 384-2815

Gabriel S. Kupchik, M.D.
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Interfaith Medical Center
Brooklyn Jewish Site
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FAX: (718) 283-6268

Ernest Lieber, M.D.
Clinical Geneticist
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FAX: (718) 857-6798

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(718) 270-2924 (Obstetrics)
FAX: (718) 270-1985

NEW YORK CITY - MANHATTAN

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Division of Medical Genetics
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APPENDIX 6:

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Clinical laboratory testing of specimens originating in New York State must be performed by appropriately licensed laboratories. The current status of a laboratory's licensure should be verified with the New York State Department of Health, Clinical Laboratory Evaluation Program, 518-485-5378 or the Genetic Testing Quality Assurance Program, 518-474-6796.

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Chantilly, VA 22021
703-802-6900

Cytogenetics - Restricted

Arleen D. Auerbach, Ph.D.
The Rockefeller University Hospital
Section of Cytogenetics
1230 York Avenue
New York, NY 10021
212-570-7533

Robert Miller, Ph.D.
Genzyme Genetics - Tampa
10770 N 46 Street, Ste A100
Tampa, FL 33617
800-966-4440

APPENDIX 7:

GENETIC TESTING LABORATORIES

Clinical laboratory testing of specimens originating in New York State must be performed by appropriately licensed laboratories. The current status of a laboratory's licensure should be verified with the New York State Department of Health, Clinical Laboratory Evaluation Program, 518-485-5378 or the Genetic Testing Quality Assurance Program, 518-474-6796.

Genetic Testing - DNA

Nathan Ellis, M.D.
Diagnostic Molecular Genetics Laboratory
Memorial Sloan-Kettering Cancer Center
1275 York Avenue
New York, NY 10021
212-639-5151

Antony Shrimpton, Ph.D.
SUNY Health Science Center
750 East Adams Street
Syracuse, NY 13210
315-464-4716

Stephen A. Brown, M.D.
DNA Diagnostic Laboratory
Columbia Presbyterian Med Ctr
630 West 168 St., Room 16-437
New York, NY 10032
212-305-6742

Arleen Auerbach, Ph.D.
The Rockefeller University Hospital
1230 York Avenue
New York, NY 10021
212-327-8862

Sylvia Spitzer, Ph.D.
University Hospital
Health Sciences Center
Stony Brook, NY 11794-7300
516-444-2600

Maria New, M.D.
Laboratory of Pediatric Endocrinology
New York Hospital
525 East 68th Street, Rm. N236
New York, NY 10021
212-746-8910

Ali Naini, Ph.D.
Molecular Neurogenetics
Columbia University
630 West 168th Street, P&S 4-448
New York, NY 10032
212-305-1541

Fred Gilbert, M.D.
DNA Diagnostic Laboratory
Cornell University School of Medicine
515 East 71 Street, Room 5-608
New York, NY 10021
212-746-3475

Howard Ratech, M.D.
Department of Pathology
Henry & Lucy Moses Division
Montefiore Medical Center
111 East 210 Street
Bronx, NY 10467
718-920-4523

Sylvia Spitzer, Ph.D.
Molecular Diagnostics Laboratory
Winthrop University Hospital
222 Station Plaza North, Suite 618
Mineola, NY 11501
516-663-4571

Jean Amos, Ph.D.
SmithKline Beecham Clinical Laboratories
7600 Tyrone Avenue
Van Nuys, CA 91405
800-877-2520

Kathryn Kronquist, Ph.D.
Specialty Laboratories, Inc.
2211 Michigan Avenue
Santa Monica, CA 90404-3900
800-421-7110

Allen E. Bale, M.D.
Department of Genetics
Yale University
333 Cedar Street
New Haven, CT 06510
203-785-5745

Stephen Gersen, Ph.D.
Dianon Systems, Inc.
200 Watson Blvd.
Stratford, CT 06497
203-381-4002

Roger Lebo, Ph.D.
Boston University Medical School
80 East Concord Street
Boston, MA 02118
617-638-7083

Bernice Allitto, Ph.D.
Genzyme Genetics
One Mountain Road
Framingham, MA 01701
800-255-7357

Gary Cutting, M.D.
DNA Diagnostic Laboratory
Johns Hopkins Hospital
600 North Wolfe St, CMSC 10-106
Baltimore, MD 21205
410-955-0483

Constance Griffin, M.D.
Pathology Molecular Diagnostics Laboratory
Johns Hopkins Medical Laboratories
600 North Wolfe Street, Oncology 109
Baltimore, MD 21287-8934
410-955-8363

Mark Rabin, Ph.D.
Gene Logic, Inc.
205 Perry Parkway
Gaithersburg, MD 20877
301-208-1888

Andrea Cortese Hassett, Ph.D.
Molecular Diagnostics Reference Laboratory
ITxM Diagnostics Inc.
3636 Boulevard of the Allies
Pittsburgh, PA 15213
412-633-7291

C. Sue Richards, Ph.D.
Baylor DNA Diagnostic Laboratory
Baylor College of Medicine
Houston, TX 77030
713-798-6536

Brian Ward, Ph.D.
Myriad Genetic Laboratories
320 Wakara Way
Salt Lake City, UT 84108
800-467-7423

Anne Maddalena, Ph.D.
Genetics & IVF Institute
3022 Javier Road
Fairfax, VA 22031
703-698-7355

Mary-Claire King, Ph.D.
King Laboratory, University of Washington
K160 Health Sciences Center
Seattle, WA 98195-7720
206-616-4294

Genetic Testing - DNA & Biochemical

Richard Erbe, M.D.
Clinical Laboratory - Genetics
Children's Hospital of Buffalo
936 Delaware Avenue
Buffalo, NY 14222
716-878-7530

Harold M. Nitowsky, M.D.
Reproductive Genetics Laboratory
Montefiore Medical Center - Einstein College
1635 Poplar Street
Bronx, NY 10461
718-405-8100

Susan Sklower-Brooks, M.D.
IBR Consolidated Clinical Laboratory
1050 Forest Hill Road
Staten Island, NY 10314
718-494-5240

Robert J. Desnick, Ph.D., M.D.
Mount Sinai Medical Center
Genetic Testing Laboratory
One Gustave Levy Place
New York, NY 10029-6574
212-241-6944

Sergio Piomelli, M.D.
Pediatric Hematology-Oncology Lab
Columbia-Presbyterian Medical Center
622 West 168th Street
New York, NY 10032
212-305-5808

Harry Ostrer, M.D.
Molecular Genetics Laboratory
New York University Medical Center
550 First Avenue
New York, NY 10016
212-263-5746

Ali Naini, Ph.D.
Clinigene Medical Specialty Laboratory
240 Motor Parkway
Hauppauge, NY 11788
800-299-3819

Edwin Kolodny, M.D.
Neurogenetics Laboratory
New York University Medical Center
400 East 34 Street, RR 213
New York, NY 10016
212-263-7637

Raymond Fenwick, Ph.D.
Quest Diagnostics Incorporated
33608 Oretga Highway
San Juan Capistrano, CA 92606-6130
714-728-4000

William Seltzer, Ph.D.
Athena Diagnostics, Inc.
377 Plantation Street
Worcester, MA 01605
800-394-4493

Stephen Thibodeau, Ph.D.
Mayo Medical Laboratories
200 First Street, SW
Rochester, MN 55905
507-284-9185

Lauren Kam-Morgan, Ph.D.
Laboratory Corporation of America Holdings
1912 Alexander Drive
Research Triangle Park, NC 27709
800-533-0567

Charles Roe, M.D.
Institute of Metabolic Disease
Baylor University
3812 Elm Street
Dallas, TX 75226
214-820-4533

Robert Montgomery, M.D.
The Blood Center of Southeastern Wisconsin
638 North 18 Street
Milwaukee, WI 53233
414-937-6250

Genetic Testing - Biochemical

Dean A. Arvan, M.D.
University of Rochester-Strong Memorial Hospital - Clinical Laboratories
601 Elmwood Avenue, Box 608
Rochester, NY 14642
716-275-3883

Peter T. Rowley, M.D.
University of Rochester School of Medicine
Division of Genetics, Box 641
601 Elmwood Avenue
Rochester, NY 14642-8641
716-275-3461

Jen C. Lin, M.D.
Nassau County Medical Center
Department of Pathology and Laboratories
2201 Hempstead Turnpike
East Meadow, NY 11554
516-542-3501

I. Herbert Scheinberg, M.D.
Wilson Disease Laboratory
432 West 58 Street, Suite 614
New York, NY 10019
212-523-8717

Maureen Poh, M.D.
St. Luke's-Roosevelt Laboratory
1000 Tenth Avenue
New York, NY 10019
212-523-8640

Frits Hommes, Ph.D.
Westchester County Medical Center
Valhalla, NY 10595
914-285-7373

Kimiyo Raymond, M.D.
Biochemical Disease Detection Laboratory
Yale University School of Medicine
333 Cedar Street
New Haven, CT 06510
203-785-2662

Marvin Natowicz, M.D.
Eunice Kennedy Shriver Center
Division of Medical Genetics
200 Trapelo Road
Waltham, MA 02254
617-642-0175

James D. Shoemaker, M.D.
Metabolic Screening Laboratory
St. Louis University
1402 South Grand
St. Louis, MO 63104
314-577-8169

Margaret McGovern, Ph.D., M.D.
Quest Diagnostics, Inc.
One Malcolm Avenue
Teterboro, NJ 07608
201-393-5000

Edwin Naylor, Ph.D.
Neo Gen Screening LP
110 Roessler Road, Suite 200D
Pittsburgh, PA 15220
412-341-8658

Stanton Segal, M.D.
Metabolic Diagnostic Laboratory
The Children's Hospital of Philadelphia
34 and Civic Center Boulevard
Philadelphia, PA 19104
215-590-3376

Edwin Naylor, Ph.D.
Biochemical Genetics Laboratory
Children's Hospital of Pittsburgh
3705 Fifth Avenue
Pittsburgh, PA 15213-2583
412-692-5778

Jerzy Tomczak, D.V.M.
Tay-Sachs Prevention Program
Thomas Jefferson University
1100 Walnut Street
MOB, Rm. 400
Philadelphia, PA 19107
215-955-8320

Appendix 8:

Descriptions of Organizations Participating in the Guidelines Project

The Alliance of Genetic Support Groups

The Alliance of Genetic Support Groups is a national coalition of voluntary genetics organizations, consumers and professionals. Serving as a bridge between people who are affected by genetic conditions and the health professionals who provide services to them, the Alliance offers a forum for addressing family needs across a spectrum of disabilities. In addition, the Alliance promotes the sharing and prompt dissemination of new information about strategies and technologies related to genetic services. They publish a newsletter and a *Directory of National Genetic Voluntary Organizations and Related Resources*. Many organizations belonging to the Alliance service and support individuals and families affected by birth defects syndromes. These include: The Support Organization for Trisomy 18/13, The Cornelia de Lange Syndrome Foundation, The National Fragile X Foundation, and hundreds of others.

The American Academy of Family Physicians

The American Academy of Family Physicians (AAFP) is a national non-profit medical association of more than 85,000 family physicians, family practice residents, and medical students. The AAFP's mission is to improve the health of patients, their families and the American people; advance and represent the specialty of family practice; and serve the unique needs of its members with professionalism and creativity. Several of the AAFP's strategic directions are relevant to this project: to encourage health promotion, disease prevention and patient education, by assuming a leadership role in improving the health of the American public; to shape public health policies through the government, public, business, and the health care industry; to assure high-quality, innovative education for practicing physicians, residents, and medical students that embodies the art, science, and socioeconomics of family medicine; and to promote new medical knowledge through primary care research, guideline development and practice.

American Academy of Pediatrics

The American Academy of Pediatrics (AAP) consists of pediatricians dedicating their efforts and resources to the health, safety and well-being of infants, children, adolescents and young adults. Activities of the AAP include advocacy for children and youth, public education, research, professional education, membership service and advocacy for pediatricians. Founded in 1930, the Academy has 53,000 members in the United States, Canada and Latin America. The AAP's Section on Genetics and Birth Defects, founded in 1990, meets for the purpose of developing ideas and programs to improve the care of infants, children and adolescents with genetic disorders by providing educational programming.

American College of Medical Genetics

In 1990, the American Society of Human Genetics Board of Directors appointed a committee to evaluate the need for and feasibility of establishing a College or Academy to oversee the

provision of genetic services nationally, to represent the providers of these services and their patients with genetic disorders, and to elevate the rapidly emerging specialty of Medical Genetics to full representation within the established medical service, certification and regulatory organizations. The committee recommended the establishment of the American College of Medical Genetics (ACMG) which was incorporated as an independent body in December of 1991. The ACMG is committed to making genetic services available to the American public without regard to age, race, national origin, gender, sexual orientation, disability or ability to pay. The College fosters the development and implementation of methods of diagnosis, treatment and prevention of genetic disease in order to improve the public health. It sponsors educational programs for geneticists, the medical community and the public at large. The College's Professional Practice and Guidelines Committee, among other activities, is responsible for the development, implementation and evaluation of clinical guidelines in medical genetics.

American College of Medical Genetics Foundation

The American College of Medical Genetics Foundation is organized exclusively for charitable, scientific, and educational purposes. More specifically, the Foundation aims to stimulate and support research, education, and knowledge in the field of medical genetics with the overall objective of advancing the art and science of medical genetics, fulfilling the medical genetic needs of the public, and, thereby, improving the public health.

American College of Obstetricians and Gynecologists

The American College of Obstetricians and Gynecologists (ACOG) is the nation's leading group of professionals providing health care for women. As a private, voluntary, nonprofit organization, ACOG serves as a strong advocate for quality health care for women; maintains the highest standards of clinical practice and continuing education of its members; promotes patient education, and stimulates patient understanding of and involvement in medical care; and increases awareness among its members and the public of the changing issues facing women's health care. Founded in Chicago in 1951, ACOG's membership has grown to more than 37,000 physicians specializing in obstetric-gynecological care.

American College of Physicians

The American College of Physicians (ACP) is the largest society of internists and allied subspecialists in the world. Founded in 1915, the ACP's mission is to enhance the quality and effectiveness of health care by fostering excellence and professionalism in the practice of medicine. ACP's goals include: to establish and promote the highest clinical standards and ethical ideals; to be the foremost comprehensive education and information resource for all internists; to advocate responsible positions on individual health and on public policy relating to health care for the benefit of the public, patients, the medical profession and ACP members; to serve the professional needs of the membership and advance internal medicine as a career; and to recognize individual excellence and distinguished contributions to internal medicine. The NYS Chapter of the ACP has 10,000 members.

American College of Radiology

The American College of Radiology (ACR) is a non-profit professional society with more than 30,000 members. The society's purposes are to advance the science of radiology, improve service to the patient, study the socio-economic aspects of the practice of radiology and encourage continuing education for radiologists and persons in allied professional fields.

American College of Surgeons

The American College of Surgeons (ACOS) was founded in 1913 to improve the standard of patient care. It is now the largest organization of surgeons in the world, with over 56,000 members. There are 97 chapters; 67 of these are in the US. A Board of Regents is elected to govern the College. Fellows have passed a rigorous evaluation process relating to education and training, professional qualifications, surgical competence, and ethical conduct. Activities of the College include sponsoring continuing medical education programs, participating in programs in patient care such as the Commission on Cancer and the Committee on Trauma, working in the legal realm through the Socioeconomic Affairs Department, and providing educational services to the public through the Office of Public Information. Monthly publications of the ACOS include the *Journal of the American College of Surgeons*, and *Bulletin*, its news magazine for fellows.

Biotechnology Industry Organization

The Biotechnology Industry Organization (BIO) is the largest trade organization to serve and represent the emerging biotechnology industry in the United States and around the globe. As the leading voice for the biotechnology industry, BIO represents 720 biotechnology companies of all sizes, academic institutions and state biotechnology centers engaged in the development of products and services in the areas of biomedicine, diagnostics, agriculture food, energy, environmental, and industrial applications. BIO is a vital forum and an invaluable focal point to bring together the resources and expertise to meet critical challenges and serve the needs of its members.

Council of Regional Networks for Genetic Services

The Council of Regional Networks for Genetic Services (CORN) was formed in 1985 to provide a forum for dialogue and national coordination among the ten regional genetics networks representing all fifty states, the District of Columbia, Puerto Rico, and the Virgin Islands. CORN brings together network representatives to encourage and facilitate communication and planning for genetics services as well as to address national public health priorities in genetics. The CORN membership includes representatives from each of the regional genetics networks, national sickle cell disease programs, and the Alliance of Genetic Support Groups, and consists of genetic services providers, public health officials, consumers and health care professionals. Goals of CORN include: to develop and maintain a uniform national system for data collection and analysis of genetic services; to develop a system of quality assurance in specialized laboratory and clinical genetic services; to plan and coordinate educational programs and information on genetics for health care professionals and consumers; to explore and advocate for methods of reimbursement for genetics services; to advocate for policies to ensure a comprehensive integrated approach to the provision of genetics services; and to

promote national efforts to improve professional and public awareness of human genetics and its impact on health and disease. CORN is a Special Project of Regional and National Significance (SPRANS), as are the ten regional networks, and is supported in part by the Maternal and Child Health Bureau, Health Resources and Services Administration, US Department of Health and Human Services.

The HMO Group

The HMO Group is a national affiliation of health maintenance organizations which were originally all physician-led staff or group model HMOs. Most are rapidly evolving to mixed model organizations. The HMO Group member plans have a combined enrollment of over 7 million members. Through The HMO Group, member plans conduct a number of activities designed to improve the process and outcome of care. Examples of collaborative activities include information sharing, bench marking, quality improvement, demonstration projects, disease management, utilization management and technology assessment.

International Society of Nurses in Genetics

The International Society of Nurses in Genetics (ISONG) is an organization which fosters scientific and professional growth of nurses in genetics. It is committed to defining the role of the genetics nurse, at both basic and advanced practice levels, and to integrating genetics into clinical practice, research and education in order to improve all aspects of genetics services. The organization has recently developed a scope and standards of practice; it is also examining opportunities for certification of nurses in genetics. As with the other professional genetics organizations, ISONG is working to promote high standards of health care and education in genetics, and is addressing the personal, ethical, legal and social issues arising from scientific and clinical developments in genetics. The activities of the American College of Medical Genetics' Clinical Guidelines Committee are consistent with ISONG's philosophy in this regard. ISONG currently has 160 members, and is involved in liaison relationships with other professional genetics and nursing organizations.

Links, Inc.

The Links, Incorporated is an organization of women comprised of more than 250 chapters and 9,500 members located throughout the United States, Germany and the Bahamas. The Links, Incorporated is a service organization of volunteers established to enhance the quality of life in the African-American community. This organization focuses on education, health and wellness, cultural enrichment and civic involvement, implemented through strategies such as public information and education, economic development, and public policy.

March of Dimes Birth Defects Foundation

The March of Dimes (MOD) is a nonprofit organization dedicated to improving the health of babies by reducing birth defects and infant mortality. The MOD is one of the ten largest voluntary health agencies in the country, with 101 chapters nationwide. The MOD offers educational programs to public and professional groups, sponsors various conferences for health care professionals throughout the country, and provides funding for research and other programs.

Medical Society of the State of New York

The Medical Society of the State of New York (MSSNY) is a federation of 61 county medical societies in NYS, and is one of the medical societies that comprise the American Medical Association. Among the purposes of the MSSNY are the following: to federate into one organization the medical profession of NYS; to contribute to the professional and personal development of member physicians by representing the profession as a whole and advocating for health-related rights, responsibilities and issues, in order to promote a favorable environment for the practice of medicine and for improvement of the health of the residents of NYS; to enhance the delivery of medical care of high quality to all people in the most economical manner; and to act to promote and maintain high standards in medical education and in the practice of medicine in an effort to ensure that quality medical care is available to the public; to seek to educate the public regarding important health care issues, including identification and discussion of their implications for patients and physicians in the current medical practice environment; and to inform and promote public opinion in regard to the problems of medicine and health in the best interests of the people of NYS.

National Society of Genetic Counselors

The National Society of Genetic Counselors (NSGC) is the leading voice and advocate for the genetic counseling profession. There are currently 1500 enrolled members of the Society; most are master's level genetic counselors. NSGC serves as a forum for discussion of professional, social and ethical issues relating to the practice of genetic counseling. It is dedicated to enhancing the quality of genetic counseling services, and to educational efforts to enable members to remain current in the field.

New York State Public Health Association

The New York State Public Health Association (NYSPHA) is an affiliate of the American Public Health Association. Its members come from all areas of Public Health practice, such as physicians, nurses, social workers, mental health workers and environmentalists. Its mission as an organization is to promote the public's health through all levels of prevention. Its members are involved with all areas of genetics practice as they care for all age groups in their home settings. The Public Health Departments in many of our counties administer the programs that care for children and adults with genetically-related conditions in their homes, at their schools, and in day care locations.

New York State Society of Internal Medicine/American Society of Internal Medicine

The New York State Society of Internal Medicine (NYSSIM) is a federation of 16 local component societies of Internal Medicine covering all of New York State. NYSSIM was incorporated in 1957 to fill a need for a dynamic organization of internal medicine specialists — vitally interested in the quality of medical care in the ever-changing social, economic and scientific aspects of internists' practice. NYSSIM is also one of the component societies of the American Society of Internal Medicine (ASIM), which represents internists in every state. ASIM is a well respected and effective voice for internal medicine in the political and socio-economic arena today. This network of local, state and national organizations represents in-

ternists' interests via a grass roots, democratic process. NYSSIM and ASIM are run by the membership. Open elections and free debate determine their policies.

The New York State Task Force on Life and the Law

Convened by the Governor in 1985, the New York State Task Force on Life and the Law has a mandate to develop public policy on a host of issues arising from medical advances, including the withholding and withdrawal of life-sustaining treatment, assisted suicide and euthanasia, assisted reproductive technologies, organ and tissue transplantation and genetic testing. Task Force members include leaders in the fields of law, medicine, nursing, philosophy and bioethics, as well as patient advocates and representatives of diverse religious communities.

Organization of Teratology Information Services (OTIS)

The Organization of Teratology Information Services is a nonprofit organization that represents organizations and individuals committed to the communication of fetal risk assessments to individuals of reproductive age and their health care providers. Members of OTIS are located throughout the United States and Canada. A collaborative relationship exists with the European Network of Teratology Information Services (ENTIS). The primary goals of OTIS are to: facilitate communication among Teratology Information Services and other organizations committed to the reduction of preventable birth defects; provide a forum for utilizing prospective pregnancy outcome data in a scientific manner; improve the quality of patient care through educational programs, an annual meeting, fact sheets, and newsletters; and maintain a mechanism for quality assurance among member programs.

Society of Craniofacial Genetics

In 1975, a group of basic scientists and clinicians interested in the developmental biology of the head and neck formed the Society of Craniofacial Genetics. As originally stated, the objectives of the Society of Craniofacial Genetics were to provide an interdisciplinary forum for those with an interest in craniofacial biology in the scientific community and the public at large. One way that the Society has striven to achieve these objectives has been to sponsor workshops and symposia on education and research, typically in collaboration with other societies and organizations.

Appendix 9:

Individuals Participating in the Guidelines Project

Members of the Steering Committee of this project provided oversight to the development, dissemination and evaluation of two clinical guidelines in medical genetics. The second guideline addressed issues in breast/ovarian cancer genetic counseling, screening and testing. Certain committee members were chosen for their expertise in oncology and related fields.

Avi Barbasch, M.D. (Steering Committee; representing American Cancer Society) is Assistant Professor of Medicine (Oncology) at the Mt. Sinai School of Medicine and Attending Physician at the Mt. Sinai Hospital in New York City. Dr. Barbasch is a member of the Board of Trustees of the American Cancer Society New York City Division and is chairman of its Program Committee. Dr. Barbasch is also a member of the Steering Committee of the Breast Cancer Guideline Development Committee of the Mt. Sinai Department of Health Policy. In addition to his large clinical practice, he is involved in clinical trials in the treatment of breast cancer.

Hope Charkins, M.S.W. (Steering Committee and Guideline Development Team; representing Alliance of Genetic Support Groups) holds a master's degree in social work from San Diego State University. She works as a medical social worker for the Vermont Department of Health, Children with Special Health Needs Program. Along with her husband, she founded the Treacher Collins Foundation, and now serves as its executive director. She has two children, one of whom has Treacher Collins syndrome. Ms. Charkins is the author of: *Children with Facial Difference: a Parent's Guide*, published by Woodbine House.

Cynthia Curry, M.D., FACMG (Steering Committee; representing American College of Medical Genetics - Professional Practice and Guidelines Committee) is Director of Genetic Medicine at Valley Children's Hospital in Fresno, California. She is a Professor of Pediatrics at the University of California, San Francisco. She received her A.B. from Mt. Holyoke College and her M.D. from Yale University. Dr. Curry has developed *Guidelines for Genetic Consultation* through the Pacific Southwest Regional Genetics Network in California and has completed a *Consensus Paper on Mental Retardation* under the auspices of the Dysmorphology Subcommittee of the Clinical Practice Committee, American College of Medical Genetics, which she chairs. Research interests include pregnancy loss, new genetic syndromes and etiologies of cerebral palsy.

Jessica G. Davis, M.D., FACMG (Steering Committee and Dissemination Team Co-Chair; representing American College of Physicians - New York State Chapter and American Public Health Association - New York State Affiliate) is the Co-Director of the Division of Human Genetics at the New York Hospital - Cornell University Medical College. Certified by the American Board of Medical Genetics in Clinical Genetics, she is an Associate Professor of Pediatrics at Cornell University Medical College. Dr. Davis is a clinical geneticist who has an active interest in genetics and public health and in genetics education for primary care providers. She is a past president of the New York State Genetics Task Force, GENES (The Genetics Network of NYS, Puerto Rico and the Virgin Islands), and CORN (The Council of Regional Networks for Genetic Services). Her clinical research interests focus on Marfan syndrome and other connective tissue disorders.

Franklin Desposito, M.D., FACMG (Steering Committee and Guideline Development Team; representing Council of Regional Networks for Genetic Services) has been Interim Chairman of the Department of Pediatrics at the University of Medicine & Dentistry of New Jersey (UMDNJ) - New Jersey Medical School since 1990. He is also Director of Clinical Genetics at the Center for Human & Molecular Genetics at UMDNJ-New Jersey Medical School. He is board certified in Clinical Genetics, Clinical Cytogenetics and Molecular Genetics. Dr. Desposito holds positions on several national committees: He is President of the Mid-Atlantic Human Regional Genetics Network (MARHGN), one of the federally funded regional genetics networks, and sits on the Steering Committee of the Council of Regional Networks for Genetics Services (CORN). Dr. Desposito has served as Chairman of the Committee on Genetics of the American Academy of Pediatrics since 1995. This committee has developed and published seven health supervision statements for children with various genetic disorders. He brings this expertise to the Guidelines project.

Charlotte Druschel, M.D., M.P.H. (Evaluation Team; New York State Congenital Malformations Registry) - Dr. Druschel is a reproductive and perinatal epidemiologist. She received her M.D. from the University of Minnesota and her M.P.H. in Epidemiology from Johns Hopkins. She has conducted extensive analyses of neonatal and postneonatal mortality in Georgia and Alabama using vital records and programmatic data. While at the Centers for Disease Control she developed a protocol for a population-based study of very low birth weight babies. While on the faculty of the School of Public Health at the University of Alabama-Birmingham, she developed courses for a new Maternal and Child Health training program. She is currently the Medical Director of the New York State Congenital Malformations Registry (CMR) where she reviews and revises coding procedures, reviews medical records and investigates potential clusters of adverse pregnancy outcomes. She oversees the state surveillance for time-space clusters and the CMR annual cohort report. She has published descriptive studies of specific malformations and has served as Co-Principal Investigator of a case-control study of heart malformations. She is the PI on three major projects: neural tube defect surveillance in the Lower Hudson Valley, the Center of Excellence for Birth Defects Prevention Research, and Fetal Alcohol Syndrome surveillance in Western New York. All of these are cooperative agreements with CDC.

Stephen B. Edge, M.D., FACS (Steering Committee; representing American College of Surgeons) is Chief of the Breast Cancer Program, Division of Surgical Oncology at Roswell Park Cancer Institute (RPCI), and Associate Professor of Surgery at SUNY at Buffalo. Dr. Edge maintains an active breast cancer practice, and serves as director of the breast cancer research development program at RPCI. He is particularly interested in breast cancer risk prediction and its clinical applications. In conjunction with Carolyn Farrell, NP, CGC who also serves on this project's Steering Committee, he is Co-Director of the Breast Cancer Risk Evaluation Program at RPCI which provides risk assessment and counseling, and offers genetic testing in appropriate circumstances for breast and ovarian cancer susceptibility genes.

Constance B. Eve, M.A. (Dissemination Team; representing Links, Inc.) is Chairperson and Founder of Women For Human Rights and Dignity, Inc., a not-for-profit human services organization whose mission is to empower women, children, and families to move on to economic, emotional, and social growth. She is a Professor Emeritus of English. She is the mother of five children and wife of Deputy Speaker Arthur O. Eve, New York State Assembly.

Among her numerous associations and affiliations are: Member, Board of Directors of the Martin House Restoration Corporation, Buffalo Chapter of Links, Alpha Kappa Alpha Sorority, and the Buffalo Council of Churches. In 1980 she founded Women For Human Rights and Dignity, Inc., which has grown to six programmatic components, and serves today as its Chairperson. For her dedication and contributions to the community at large she has received countless awards, honors, and citations, among which are: Outstanding Citizen of New York State Award from Board of Trustees, State University of New York, Albany, New York; Citizen of the Year, Buffalo Evening News and 1997 YWCA of Western New York Leader Award.

Carolyn D. Farrell, M.S., C.N.P., CGC (Steering Committee; representing International Society of Nurses in Genetics) is Director of Clinical Genetic Services at Roswell Park Cancer Institute. She is a Diplomate of the American Board of Medical Genetics, a Board Certified Genetic Counselor, a Board Certified Nurse Practitioner, and has had 15 years of experience in genetics. She is an active member of several professional genetics societies and oncology organizations, including the American Society of Human Genetics, American College of Medical Genetics, National Society of Genetic Counselors, International Society of Nurses in Genetics (President), Oncology Nursing Society, and others. Ms. Farrell has lectured extensively and has published on various genetics topics, and the related ethical, legal, social and professional issues. She is co-director and developer of a comprehensive program for evaluation, education and testing of persons with an individual or family history of breast or ovarian cancer. She has received a National Cancer Institute Supplemental Grant for Education and Training in Cancer and Genetics; this is focused on three target groups - physicians, allied health care professionals, and the lay public.

Beth A. Fine, M.S., CGC (Steering Committee; represented American College of Medical Genetics - Joint Committee on Professional Practice and Guidelines) was an Assistant Professor of Obstetrics and Gynecology and Coordinator of the Graduate Program in Genetic Counseling at Northwestern University Medical School in Chicago. Over the past 18 years, she practiced genetic counseling in pediatric, prenatal and cancer genetics clinics. Ms. Fine was a past president of the National Society of Genetic Counselors and was Vice President of the American Board of Genetic Counseling. She served as a member of the Executive Committee of the ELSI Working Group, as an advisory panel member for the Office of Technology Assessment's study, *Cystic Fibrosis and DNA Tests: Implications for Carrier Screening*, and as a member of the Expert Panel for the NIH Technology Conference on *Gaucher Disease: Current Issues in Diagnosis and Treatment*. These activities and her work in establishing a Breast Cancer Risk Assessment and Genetic Counseling Program at the Lynn Sage Breast Cancer Center at Northwestern Memorial Hospital informed her contributions to the development of these Guidelines.

Alan R. Fleischman, M.D. (Steering Committee and Evaluation Team; representing New York State Task Force on Life and the Law) is Senior Vice President of the New York Academy of Medicine and Clinical Professor of Pediatrics and Clinical Professor of Epidemiology and Social Medicine at the Albert Einstein College of Medicine in New York City. He graduated from the City College of New York and from the Albert Einstein College of Medicine. He continued his education in Pediatrics and Neonatology at the Johns Hopkins Hospital and at the National Institutes of Health. Dr. Fleischman joined the faculty at the Albert Einstein

College of Medicine and the Montefiore Medical Center in 1975, where he became Director of the Division of Neonatology and Professor of Pediatrics and Professor of Epidemiology and Social Medicine. In 1994, he became Senior Vice President of the New York Academy of Medicine, where he is responsible for initiatives in urban health, education, public policy, ethics and public health. He has published over 100 articles and book chapters in the areas of perinatal medicine and bioethics.

Lizbeth M. Fonseca, M.S. (Project Coordinator) is Coordinator of the Clinical Guidelines Project for the American College of Medical Genetics. The College has developed two sets of clinical practice guidelines for healthcare providers. The first guideline concerns breast/ovarian cancer genetic screening, counseling and testing; the second focuses on diagnosis, management and genetic counseling for families of newborns with single or multiple congenital anomalies. Ms. Fonseca is Health Program Administrator for the New York State Department of Health's Genetic Services Program in the Wadsworth Center. She joined the program after receiving her degree in Health Policy and Management from the State University of New York's School of Public Health. She serves on the Advisory Board of the Upstate Cancer Genetics Screening Initiative (Jenilu Project) based at the Saratoga Institute. She is particularly interested in social, ethical and legal issues relating to health care and in the implications of bio-technologies involving genetics.

Paul Gitman, M.D., FACP (Steering Committee; representing American Society of Internal Medicine) is a practicing General Internist and Vice President of Clinical Care and Resource Management at Long Island Jewish Medical Center. He is board certified in Internal Medicine and is an Assistant Professor of Medicine at Albert Einstein College of Medicine. Active in numerous medical organizations, Dr. Gitman is immediate past president of the New York State Society of Internal Medicine. He is Chair of the Health and Public Policy Committee and a member of the Governing Council of the New York State Chapter of the American College of Physicians. He is Chair of the Island Peer Review Organization's Criteria Norms and Standards Committee and is a member of the Steering Committee of their Health Care Quality Improvement Program. Dr. Gitman is the elected Chair of the New York State Carrier's Advisory Committee and is Chair of the Organized Medical Staff Section of the Medical Society of the State of New York. Dr. Gitman has also served as a delegate and chairman of the New York State Delegation to the AMA's Organized Medical Staff Section where he was responsible for the submission of numerous resolutions to the AMA. Dr. Gitman was appointed by the Governor to serve on a panel to review the New York State Physician Discipline Process and in June of 1997 was appointed to the New York State Board of Medicine.

Rosalie B. Goldberg, M.S., CGC (Steering Committee and Guideline Development Team; representing National Society of Genetic Counselors) is the Genetic Counselor for the Family Core of the Human Genetics Program at the Albert Einstein College of Medicine (AECOM), Bronx, New York, and Acting Director of the Velo-Cardio-Facial Institute at Montefiore Medical Center. She is a Senior Associate on the faculties of Plastic Surgery, Pediatrics and Molecular Genetics at the AECOM and is the coordinator of molecular genetic studies as correlated with clinical findings in congenital craniofacial disorders, especially velo-cardio-facial syndrome (22q11 deletion syndrome). She has co-authored numerous peer-reviewed articles on the genetic, physical and psychosocial impact of craniofacial disorders and has a number of disorders that are named after her. Ms. Goldberg serves as the National Society of Genetic Coun-

selors (NSGC) liaison to the National Human Genome Research Institute (NHGRI) Advisory Council. She has been active in the NSGC since it was incorporated in 1979 and in 1994 received an NSGC Regional Leadership Award.

Karen Greendale, M.A., CGC (Co-Principal Investigator) is currently working in the Bureau of Chronic Disease Services, Division of Chronic Disease Prevention and Adult Health, New York State Department of Health. She previously served as Genetic Services Program Administrator in the Wadsworth Center, New York State Department of Health and was co-coordinator of the Genetics Network of New York State, Puerto Rico and the Virgin Islands. Ms. Greendale is a genetic counselor certified by the American College of Medical Genetics in 1982 and by the American Board of Genetic Counseling in 1993. She is a past president of the National Society of Genetic Counselors and chaired the Council of Medical Genetics Organizations in 1994. She is a member of the American College of Medical Genetics Professional Practice and Guidelines Committee and acts as liaison to the College from the National Society of Genetic Counselors. Her research interests are in quality improvement in genetic counseling and genetic service delivery, genetics health workforce issues, international provision of genetic services, and cancer genetics.

Kurt Hirschhorn, M.D., FACMG (Steering Committee; representing American College of Medical Genetics Foundation) received his B.A. in 1950 from New York University and his M.D. degree in 1954 from New York University School of Medicine. After a residency in internal medicine and fellowship at Bellevue Hospital in New York City, he completed a fellowship in Human Genetics in Uppsala, Sweden. He became Assistant Professor and then Associate Professor of Medicine at New York University School of Medicine. He became Professor of Pediatrics at Mount Sinai School of Medicine in 1966 where he established a new Medical Genetics Program. From 1968 to 1976 he was the Arthur J. and Nellie Z. Cohen Professor of Genetics at Mount Sinai School of Medicine. In 1977, he became the Herbert H. Lehman Professor and Chairman of the Department of Pediatrics at Mount Sinai School of Medicine and Pediatrician-in-Chief at Mount Sinai Hospital. He is now Professor of Pediatrics, Human Genetics and Medicine at Mount Sinai School of Medicine. He has worked in cytogenetics, biochemical genetics, immunogenetics and molecular genetics, publishing over 300 papers and reviews. In the early 1960s, he discovered the mixed lymphocyte reaction which was one of the major beginnings of the field of cellular immunology and immunogenetics. He made fundamental contributions in the area of inborn errors of metabolism, including several lysosomal enzyme defects and Menkes syndrome. Dr. Hirschhorn is a member of many societies, including the American Society of Human Genetics (President, 1969), Harvey Society (President, 1980), American Society for Clinical Investigation, Association of American Physicians, American Pediatric Society, American Academy of Pediatrics (Fellow), American College of Medical Genetics (Honorary and Founding Fellow), the AAAS (Fellow) and the Institute of Medicine of the National Academy of Sciences (Senior Member). He is one of the founding members of The Hastings Center for Biomedical Ethics. He received the William Allan Award of the American Society of Human Genetics in 1995. He has served or is currently serving on many Editorial Boards. He has served as a Consultant or Member of the Advisory Board of the NIH, National Foundation-March of Dimes Basic Sciences Research Committee (Chairman), National Tay-Sachs Association, Dysautonomia Foundation (Chairman), American Board of Medical Genetics (Founding Member) and American Cancer Society (Council).

Michael M. Kaback, M.D., FACMG (Principal Investigator) is a Professor in the Departments of Pediatrics and Reproductive Medicine, and Chief, Division of Medical Genetics, Department of Pediatrics, University of California, San Diego (UCSD). He is the Director of the State of California Tay-Sachs Disease Prevention Program and of the International Tay-Sachs Disease Testing, Quality Control, and Data Collection Program. He received a B.A. from Haverford College and an M.D. from the University of Pennsylvania. After training and fellowships (Johns Hopkins and National Institututes of Health) he has held faculty positions at Johns Hopkins, University of California at Los Angeles and UCSD. From 1986-1992, he served as Chair, Department of Pediatrics, UCSD, and Pediatrician-in-Chief, Children's Hospital & Health Center, San Diego. Dr. Kaback is a Founding Board Member of the American College of Medical Genetics and chairs its Professional Practice and Guidelines Committee. He has served as Vice President of the Society for Pediatric Research, President of the Western Society for Pediatric Research and President of the American Society of Human Genetics, and is currently President-elect of the American Pediatric Society. Dr. Kaback is a member of the Institute of Medicine, National Academy of Sciences, and a Fellow of the American Association for the Advancement of Science. He is a recipient of the 1993 William Allan Award from the American Society of Human Genetics, for outstanding contributions to human genetics. Dr. Kaback is the North American Editor for *Prenatal Diagnosis* and Associate Editor for *Advances in Pediatrics.* He has authored or co-authored eight books and more than 200 publications in the biomedical literature. Dr. Kaback's major research interests include genotype-phenotype correlations in the lysosomal storage disorders, applications of genetic technologies to the control of human genetic disease, and psychosocial issues in population-based genetic screening.

Elly Kirschner, M.A. (Steering Committee; representing National Alliance of Breast Cancer Organizations) is Manager of Information Services for the National Alliance of Breast Cancer Organizations (NABCO), the leading non-profit central information resource on breast cancer and a network of over 375 breast cancer organizations. Ms. Kirschner manages a trained staff who provide breast cancer information to patients and their families, health professionals, and the media. Ms. Kirschner oversees NABCO's Web Site, develops educational materials, manages educational programs and projects, and represents NABCO and its interests at meetings and on committees, most recently at the American Society of Clinical Oncologists Train-the-Trainer Workshop on Cancer Genetics Education as a panel member representing the consumer perspective on genetic testing. Ms. Kirschner has been involved in the public health field for eight years. Most recently, she has been working on her Doctor of Science degree at the Harvard School of Public Health. Prior to her studies, Ms. Kirschner was Curriculum Coordinator for the *Know Your Body* comprehensive school health program at the American Health Foundation.

Dale Halsey Lea, R.N., M.P.H., CGC (Evaluation Team Chair; representing International Society of Nurses in Genetics) has been a practicing nurse for twenty years, and a Board-certified genetic counselor for the past ten years at the Foundation for Blood Research. She is the Senior Genetic Counselor and Supervisor of the Clinical Genetics Division, Southern Maine Regional Genetics Service. Ms. Lea holds a baccalaureate degree in nursing from Westbrook College, Portland, Maine and a Masters in Public Health from Loma Linda University, Loma Linda, California, with a focus on health education and health promotion.

Primary areas of genetic counseling and nursing practice include pediatric and general genetics, prenatal and specialty clinics (hemophilia and cleft lip/palate). Areas of interest include quality improvement in genetic counseling services, and promotion of education in genetics for all nurses. Ms. Lea has published widely in the nursing literature on genetics topics for nurses, and is the first author of the book Genetics and Nursing: New Directions for Clinical Practice, published by Jones and Bartlett Publishers. Ms. Lea is a past president of the International Society of Nurses in Genetics, Inc. (ISONG), and has served as chair of the following ISONG committees: Bylaws, Education, and Ethics and Social Policy. She was Chair of the 1997 Annual ISONG Education Program. A major interest and focus of Ms. Lea's career has been the support of collaboration with primary health care professionals to provide quality genetics services to patients and families with genetic concerns.

Lauren Lynch, M.D., FACMG (Steering Committee and Guideline Development Team; representing American College of Obstetricians and Gynecologists) is an obstetrician with subspecialties in Maternal Fetal Medicine and Clinical Genetics. Dr. Lynch is certified in Obstetrics and Gynecology and Maternal Fetal Medicine by the American Board of Obstetrics and Gynecology. She is also certified in Clinical Genetics by the American Board of Medical Genetics. Dr. Lynch has a special interest in fetal diagnosis and therapy and most of her research experience is in these areas.

Mary Louise F. Merola, M.D. (Steering Committee and Guideline Development Team; representing American Academy of Family Physicians) is a board certified family physician who has been active in the New York State Academy of Medicine since her residency. She has served on various committees to assist in patient education in the areas of domestic violence, breast cancer and health maintenance. She is in private practice and volunteers her time at a community hospital based residency program. She was requested by the state and national academy to represent primary care physicians on the committee. She has served on the New Physicians Committee of the American Academy of Family Physicians, has chaired the Education Committee of the New York State Academy of Family Physicians, and is presently on the Board of Directors of the New York State Academy of Family Physicians.

Kenneth Offit, M.D., M.P.H., FACMG (Steering Committee) is Chief of the Clinical Genetics Service in the Department of Human Genetics at Memorial Sloan-Kettering Cancer Center. He was Chair of the subcommittee on Cancer Genetic Testing of the American Society of Clinical Oncology and headed an advisory panel to the National Cancer Institute Director that outlined potential directions for a National Cancer Genetics Network. Dr. Offit's team identified the most frequent breast cancer susceptibility mutation occurring in individuals of Ashkenazi Jewish background. His current research focuses on other founder mutations in cancer susceptibility syndromes, and on the psychological, ethical, and policy implications of cancer genetics predisposition testing. He has been interested in the interplay between genetic and environmental causes of cancer, and has developed a clinical program to counsel individuals at hereditary risk for cancer. The goal of these efforts is to prevent cancers with an inherited susceptibility or to detect them at the earliest, most curable stage. Since its inception, the Clinical Genetics Service has screened the family histories of over 6,000 individuals with cancer, and it has helped more than 1,000 families identify a familial risk for cancer. Research programs focus on genotype-phenotype correlations of breast/ovarian can-

cer susceptibility genes, and the impact of counseling. There is also a special research focus on founder mutations of BRCA and HNPCC genes, and on familial lymphoma.

Stirling M. Puck, M.D., FACMG (Steering Committee; representing Biotechnology Industry Organization) received her B.A. from Harvard University in 1969 and her M.D. from Yale University in 1975. She remained at Yale to complete her pediatric training in 1978 and her fellowship in Human Genetics in 1980. She is certified by the American Board of Pediatrics and by the American Board of Medical Genetics in Clinical Genetics and Clinical Cytogenetics. She is a member of the American Society of Human Genetics and is a founding fellow of the American College of Medical Genetics. Since 1980 she has lived in Santa Fe, New Mexico, where she is a genetics consultant to the State Health Department and a member of the State Committee on Newborn Screening. She pioneered the private practice of genetics in both the clinical and laboratory setting. She became Medical Director of Vivigen, Inc., in 1982 and continues in that position today for Genzyme Genetics. Working through the Mountain States Regional Genetics Services Network, Dr. Puck has been instrumental in establishing maternal serum screening for open neural tube defects and Down syndrome as standard of care. As Medical Director of one of the largest genetics organizations in the world, she is responsible for maintaining the high quality and effective delivery of the clinical, cytogenetic, alpha fetoprotein screening and molecular diagnostic services the network provides. She also determines when new technologies are suitable for incorporation into Genzyme Genetics' clinical or laboratory practice, and writes educational materials for patients, physicians and other health care professionals.

Cheryl S. Reid, M.D., FAAP, FACMG (Guideline Development Team Co-Chair; representing Society of Craniofacial Genetics) is a pediatrician and medical geneticist. Prior to entering private practice, she was head of the Genetics Division at Cooper Hospital/University Medical Center in Camden, NJ, Director of the Southern New Jersey Regional Genetics Center and a faculty member at UMDNJ - Robert Wood Johnson Medical School, achieving the rank of Associate Professor of Clinical Pediatrics and Clinical Associate Professor of Obstetrics and Gynecology. She is Board certified in both Pediatrics and Medical Genetics, Clinical Genetics. In addition to patient care, she consults for health care organizations interested in genetics issues and health care for children and is an Adjunct Professor in Genetic Counseling at Beaver College in Glenside, PA. Dr. Reid has served on committees for numerous organizations, including the March of Dimes, the American Cleft Palate Craniofacial Association, the Cleft Palate Foundation, the American Academy of Pediatrics and its Section on Genetics and Birth Defects, the American College of Medical Genetics, the Human Genetics Association of New Jersey and the Society of Craniofacial Genetics, for which she was president in 1994-96. She has also been chosen for honorary membership by the Little People of America, Inc., a patient organization for individuals with short stature. She is the Genetics Section Editor for the *Cleft Palate Craniofacial Journal* and has made several recent presentations on genetics and specialty services in managed care.

John D'A. Reinhard, M.D. (Steering Committee; representing The HMO Group) is responsible for the technology assessment (TEMINEX) program of the HMO Group. Dr. Reinhard is an Associate Clinical Professor in the Department of Medicine at the State University of New York at Buffalo School of Medicine. He is a Senior Physician Manager at Health Care Plan, a mixed model HMO in Buffalo, New York. His responsibilities include Case Manage-

ment, Technology Assessment and medical support for Benefit Design and Interpretation. He is also a practicing general Internist and Rheumatology consultant at Health Care Plan. He is a Section Editor of *HMO Practice*, the journal of the HMO Group. He has also become active at the national level in exploring how academic medical centers can work with managed care organizations to improve the processes and outcomes of care, utilizing the unique strengths and resources of each type of organization.

Luther Robinson, M.D. (Steering Committee and Guideline Development Team Co-Chair; representing Organization of Teratology Information Services) - Luther K. Robinson received the MD degree from the University of Cincinnati School of Medicine and completed a postdoctoral fellowship in Dysmorphology at the University of California, San Diego Medical Center. Dr. Robinson is certified by the American Board of Medical Genetics in Clinical Genetics. He is currently an Associate Professor in the Department of Pediatrics at the State University of New York at Buffalo School of Medicine and Biomedical Sciences and is Director of Clinical Genetics in the Division of Genetics at Children's Hospital of Buffalo. Dr. Robinson is also Medical Director of the Western New York Teratology Information Service and represents the Genetics Network of New York State, Puerto Rico and the Virgin Islands on the Council of Regional Genetics Networks Committee on Teratogen Information Services. Dr. Robinson's research interests include evaluations of infants with prenatal exposures to suspected teratogens, delineation of new malformation syndromes, and new approaches to prevention education.

Ruth Rosenblatt, M.D., FACR (Steering Committee; representing American College of Radiology) is Director of Women's Imaging, Department of Radiology of New York Hospital/Cornell Medical Center. She has been a member of the American College of Radiology for over 20 years and a fellow since 1984. Dr. Rosenblatt received her medical degree from the Medical College of Pennsylvania and specialty training in radiology at Montefiore Medical Center where she was also on staff for over 20 years. Her major clinical interests have been in detection of breast cancer and gynecological disease using mammography and ultrasound respectively. She has written book chapters, published articles and lectured widely on ultrasound and mammography. Dr. Rosenblatt is past president of the New York Roentgen Society and immediate past president of the New York State Radiological Society, a chapter of the American College of Radiology.

Lawrence R. Shapiro, M.D., FACMG (Steering Committee and Guideline Development Team; representing American Academy of Pediatrics, March of Dimes Birth Defects Foundation and Medical Society of the State of NY) represents the American Academy of Pediatrics and The March of Dimes Birth Defects Foundation, of which he has been a grant recipient for clinical genetics services for many years. He also represents the Medical Society of the State of New York. He is currently Professor of Pediatrics and Pathology at New York Medical College and Director of the Regional Medical Genetics Center at Westchester County Medical Center in Valhalla, New York. He is Board Certified by the American Board of Medical Genetics in both Clinical Genetics and Clinical Cytogenetics and by the American Board of Pediatrics. He is a Founding Fellow of the American College of Medical Genetics and is a Fellow of the American Academy of Pediatrics and Board Member of the Section on Genetics and Birth Defects. Dr. Shapiro's interests involve the delineation and understanding of congenital malformations, mental retardation, cytogenetics and prenatal diagnosis. His most recent research interests have dealt with the Fragile X syndrome.

Stephen J. Shochat, M.D. (Guideline Development Team; representing American College of Surgeons) is Surgeon-in-Chief and Chairman of the Department of Surgery at St. Jude Children's Research Hospital and Professor of Surgery and Pediatrics at the University of Tennessee Health Science Center. He was previously a senior surgeon at the Children's Hospital of Philadelphia (1994-1996) and Chief of the Division of Pediatric Surgery and Professor of Surgery and Pediatrics at Stanford University School of Medicine (1977-1994). He represents the American College of Surgeons. He is interested in the surgical management of neonates with multiple congenital anomalies and has published a number of papers concerning neonatal surgery, specifically the management of congenital diaphragmatic hernia.

Jeffery P. Struewing, M.D., M.S. (Steering Committee) received his medical degree from the Indiana University School of Medicine in 1985 and an M.S. in Preventive Medicine from the University of Maryland in 1988. He is a Senior Research Investigator at the Genetic Epidemiology Branch, National Cancer Institute, Bethesda, Maryland *[Dr. Struewing is not representing any federal agency in his work on the Clinical Guidelines project]*. Dr. Struewing's research interests focus on breast and ovarian cancer genetic epidemiology, molecular genetics, and policy.

Cherie Trabert, M.S., R.N., P.N.P. (Steering Committee; representing the American Public Health Association - New York State Affiliate) is a pediatric practitioner whose practice specialty and setting is Public Health. She has directed public health programs in rural, HMO and proprietary home health agencies. In these capacities she has been intimately involved in the care of children with genetic disorders in the home setting. She has orchestrated new programming for these children through special grants. She has also participated in setting up breast cancer screening clinics in a rural area.

Laura Zajac, M.S., CGC (Dissemination Team Co-Chair; representing National Society of Genetic Counselors) received a Master's degree in genetic counseling from Northwestern University in Chicago in 1992. She is certified by the American Board of Medical Genetics, subspecialty in genetic counseling, and became a charter member of the American Board of Genetic Counseling in 1993. She has been employed as a genetic counselor in the Human Genetics Program at New York University Medical Center since 1992. She was appointed Clinical Instructor of Pediatrics at NYU Medical Center in 1996. Her professional activities include prenatal and pediatric counseling with a special interest in counseling patients with abnormal ultrasound findings and dysmorphology. She teaches medical students and pediatric and obstetric residents in classroom and clinical settings. She participates in multidisciplinary conferences and case management teams for appropriate patient diagnosis and ongoing care.

Figure 2: Components of Evaluation and Care Reference Card

Components of Evaluation and Care Reference Card
American College of Medical Genetics

Evaluation of the Newborn with Malformations

Components of Evaluation and Care

History

- Prenatal
  - Maternal age, parity, health
  - Complications of pregnancy
  - Illnesses and treatments
  - Exposures (drugs, etc)
  - Folic acid supplementation
  - Prenatal testing
- Perinatal
  - Duration of pregnancy
  - Intrapartum course
  - Presentation and mode of delivery
  - Complications and course
- Family history
  - Information re: three generations
  - Malformed infants
  - Stillborns
  - Familial disorders or traits
  - Consanguinity
  - Ethnic background

Physical Examination

- Gestational age, growth parameters and measurements (%iles)
- Comprehensive examination
  - General appearance
    - Proportionality and symmetry
    - Positioning, posture
    - Behavior
    - Other observations
  - Skin pigmentation, dimples, peeling, vascular or other lesions
  - Head shape, symmetry, fontanelles
  - Scalp hair patterning and whorls
  - Facial features
    - Pupils, orbits, palpebral fissure length and slant
    - Ear rotation, size and shape
    - Nasal appearance and patency
    - Lips, philtrum and vermilion border
    - Palate, tongue and alveolar ridges
    - Mandibular shape and size
  - Nuchal posterior hairline, redundant skin or webbing, sinus tracts
  - Chest shape, symmetry, location of nipples, +/- accessory nipples
  - Heart murmurs and pulses
  - Lung sound symmetry

Components of Evaluation and Care Reference Card
New York State Department of Health

Physical Examination (continued)

- Abdominal wall and palpation abnormalities, e.g. masses
- Genital appearance and maturation
- Anal orifice location and patency
- Back symmetry +/- sinus tracts or hair tufts in intergluteal cleft
- Extremity proportion, appearance, range of motion, abnormalities
  - Palmar creases, flexion creases at joints
- Neurological exam, including tone, alertness & reflexes

Initial Impression & Differential Diagnosis

- Single (isolated) malformation
- Multiple malformations of known pattern (syndrome identified)
- Multiple malformations (pattern not recognized)

Diagnostic Evaluations

- Imaging
- Chromosomal analysis
- Other genetic tests
- Consider specialty referral and/or consultation

Develop Working Diagnosis and Counsel Family

Counseling the Family

- Counseling principles
  - Supportive
  - Respect privacy and confidentiality
  - Facilitate grieving process
- Supportive setting
  - Privacy and quiet
  - Inclusion of others according to family preference
- Content
  - Medical facts and diagnoses
  - Etiology and prognosis
  - Treatment plan and priorities
  - Uncertainties
  - Resources
  - Recurrence risks
  - Family psychosocial support

Longitudinal Care and Case Management

- Written reports and written materials for families
- Ongoing care and referrals as indicated